LA PAROLA del22

Roma, 24-25 Maggio 2013

DIAGNOSI E TERAPIA IN ITALIA

Bruno Marino

Department of Pediatrics
Sapienza University of Rome
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La delezione 22: le pietre miliari

- 1965 – DiGeorge Syndrome

- 1976 – ConoTruncal Anomaly Face

- 1978 – Velocardiofacial Syndrome

- 1981 –

A Deletion in Chromosome 22 Can Cause DiGeorge Syndrome

Albert de la Chapelle[1], Riitta Herva[2], Maila Koivisto[3], and Pertti Aula[4]

Hum Genet (1981) 57:253–256

THYMOPOIETIN PENTAPEPTIDE TREATMENT OF PRIMARY IMMUNODEFICIENCIES

F. Aiuti, M. Fiorilli, I. Quinti, R. Seminara, L. Businco, E. Galli, P. Rossi, G. Goldstein

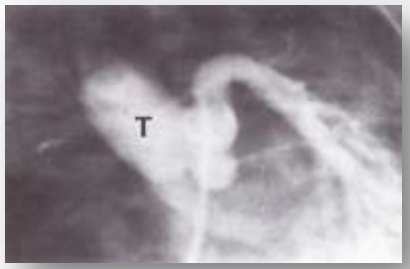
- Departments of Clinical Immunology and Paediatrics, and Institute of Internal Medicine IIIrd, University of Rome, United Kingdom
- Ortho Pharmaceutical Corporation Raritan, New Jersey, U.S.A..

Lancet 1983; 321: 551-5

DIGEORGE ANOMALY ASSOCIATED WITH PARTIAL DELETION OF CHROMOSOME 22

Report of a case with X/22 translocation and review of the literature

B. DALLAPICCOLA, B. MARINO, A. GIANNOTTI, Giuditta VALORANI



Ann Génét 1989;32:92-96

DALLAPICCOLA B., MARINO B., GIANNOTTI A., VALORANI Giuditta. — DiGeorge anomaly associated with partial deletion of chromosome 22. Report of a case with X/22 translocation and review of the literature.
Ann Génét, 1989, **32,** nº 2, 92-96.

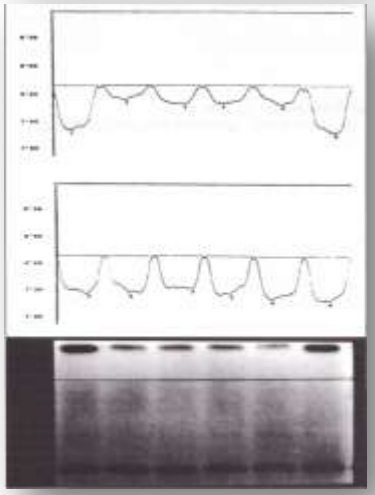
SUMMARY : Partial monosomy of 22q, resulting from a de novo unbalanced translocation t(X;22)(q28;q11) was detected in a newborn female with manifestations of the DiGeorge anomaly including multiple anomalies, type I truncus arteriosus, T-cell abnormalities, thymic aplasia and parathyroid hypoplasia noted on postmortem examination. Although DiGeorge anomaly is causally heterogeneous, our patient, together with 18 previously known cases, confirm that partial monosomy of the proximal long arm of chromosome 22 is the single most common cause of this polytopic developmental field defect.

KEY-WORDS : DiGeorge anomaly. — Chromosome translocation del(22)(q11) X chromosome. — Thymic aplasia. — Developmental field defect. — Neurocristopathy.

Delezione 22q11.2

- **Diagnosi clinica**

- **Diagnosi genetica**



Del22: contributi clinici - 1

Cayler Cardiofacial Syndrome and del 22q11: Part of the CATCH22 Phenotype

American Journal of Medical Genetics 53:303–304 (1994)

Aldo Giannotti
Maria Cristina Digilio
Bruno Marino
Rita Mingarelli
Bruno Dallapiccola

Am J Med Genet 1994; 53: 303-304

Del22: contributi clinici - 2

Maternal Diabetes Causing DiGeorge Anomaly and Renal Agenesis

American Journal of Medical Genetics 55:513–514 (1995)

Maria Cristina Digilio
Bruno Marino
Roberto Formigari
Aldo Giannotti

Am J Med Genet 1995; 55: 513-514

Del22: contributi clinici - 3

Radial aplasia and chromosome 22q11 deletion

Maria Cristina Digilio, Aldo Giannotti, Bruno Marino, Anna Maria Guadagni, Marcello Orzalesi, Bruno Dallapiccola

J Med Genet 1997; 34: 942-944

Del22: contributi clinici - 4

Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study

A K Ryan, J A Goodship, D I Wilson, N Philip, A Levy, H Seidel, S Schuffenhauer, H Oechsler, B Belohradsky, M Prieur, A Aurias, F L Raymond, J Clayton-Smith, E Hatchwell, C McKeown, F A Beemer, B Dallapiccola, G Novelli, J A Hurst, J Ignatius, A J Green, R M Winter, L Brueton, K Brøndum-Nielsen, F Stewart, T Van Essen, M Patton, J Paterson, P J Scambler

J Med Genet 1997; 34: 798-804

Del22: contributi clinici - 5

Microdeletion 22q11 and oesophageal atresia

Maria Cristina Digilio, Bruno Marino, Pietro Bagolan, Aldo Giannotti, Bruno Dallapiccola

J Med Genet 1999; 36: 137-9

Del22: contributi clinici - 6

Audiological findings in patients with microdeletion 22q11 (di George/velocardiofacial syndrome)

M.C. Digilio[1], C. Pacifico[2], L. Tieri[2], B. Marino[3], A. Giannotti[1] and B. Dallapiccola[4]

[1]Departments of Medical Genetics, [2] Audiology and [3] Pediatric Cardiology, Bambino Gesù Hospital, Rome, Italy and Department of Genetics, Tor Vergata University and Mendel-CSS Institute, Rome, Italy

Br J Audiol 1999; 33: 329-334

Del22: contributi clinici - 7

Auxological evaluation in patients with DiGeorge/velocardiofacial syndrome (deletion 22q11.2 syndrome)

Maria Cristina Digilio, MD[1] , Bruno Marino, MD[2], Marco Cappa, MD[3], Paola Cambiaso, MD[4], Aldo Giannotti, MD[1], and Bruno Dallapiccola, MD[5]

Genet Med 2001; 3: 30-3

Del22: contributi clinici - 8

Screening for Celiac Disease in Patients With Deletion 22q11.2 (DiGeorge/Velo-Cardio-Facial Syndrome)

Maria Cristina Digilio*
Aldo Giannotti
Massimo Castro
Franco Colistro
Francesca Ferretti
Bruno Marino
Bruno Dallapiccola

Am J Med Genet 2003; 121A: 286-8

Del22: contributi clinici - 9

Postural anomaly of the head-neck-shoulder alignment in patients with deletion 22q11.2 (DiGeorge/velocardiofacial syndrome)

MC Digilio
A Giannotti
B Dallapiccola
B Marino

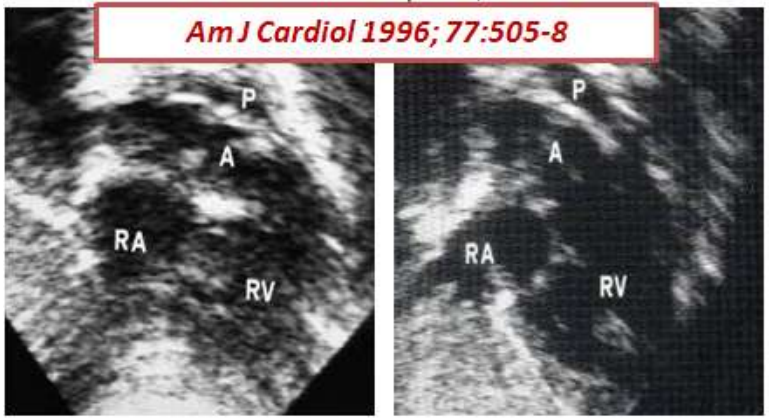
Clin Genet 2003; 64: 447-8

Del22: contributi cardiologici

Associated Cardiac Anomalies in Isolated and Syndromic Patients With Tetralogy of Fallot

Bruno Marino, MD, Maria Cristina Digilio, MD, Sabina Grazioli, MD, Roberto Formigari, MD, Rita Mingarelli, MD, Aldo Giannotti, MD, and Bruno Dallapiccola, MD

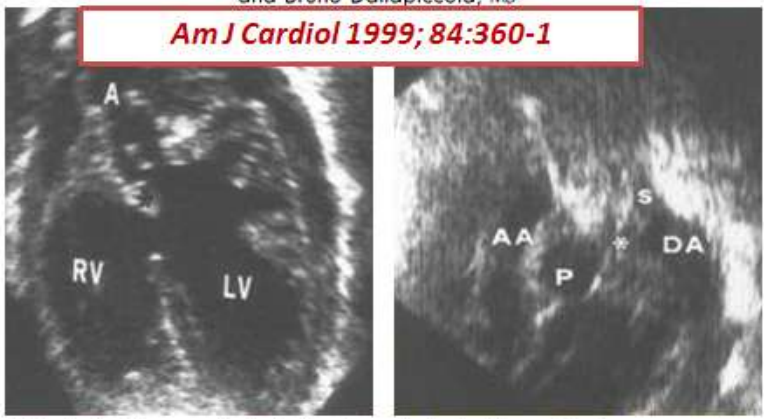
Am J Cardiol 1996; 77:505-8



Deletion 22q11 in Patients With Interrupted Aortic Arch

Bruno Marino, MD, Maria Cristina Digilio, MD, Monica Persiani, MD, Roberto Di Donato, MD, Alessandra Toscano, MD, Aldo Giannotti, MD, and Bruno Dallapiccola, MD

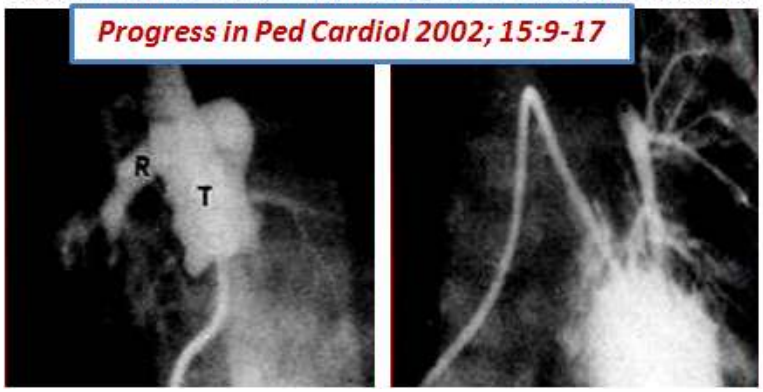
Am J Cardiol 1999; 84:360-1



Common arterial trunk, DiGeorge syndrome and microdeletion 22q11

Bruno Marino*, M. Cristina Digilio, Alessandra Toscano

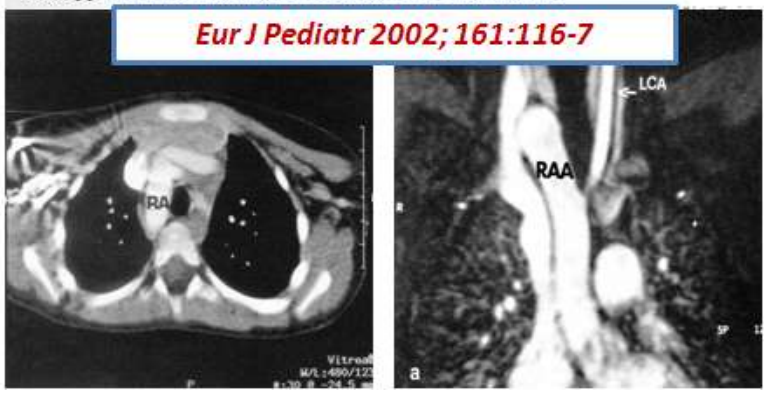
Progress in Ped Cardiol 2002; 15:9-17



Ventricular septal defect and deletion of chromosome 22q11: anatomical types and aortic arch anomalies

Alessandra Toscano · Silvia Anaclerio
Maria Cristina Digilio · Aldo Giannotti
Giuseppe Fariello · Bruno Dallapiccola · Bruno Marino

Eur J Pediatr 2002; 161:116-7

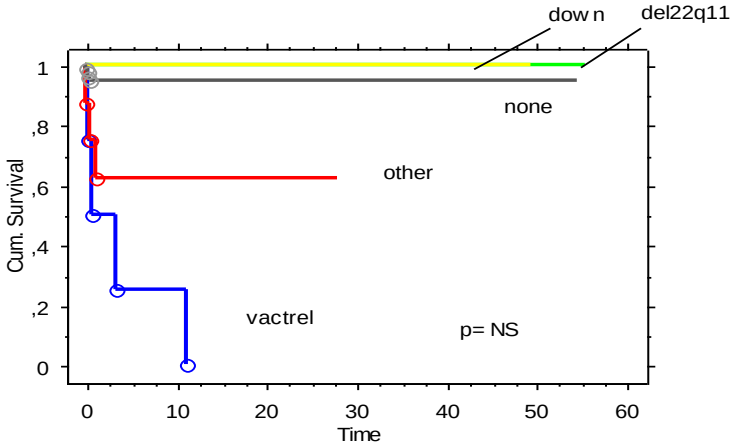


Del22: risultati cardiochirurgici

Genetic Syndromes and Outcome After Surgical Correction of Tetralogy of Fallot

Guido Michielon, MD, Bruno Marino, MD, Roberto Formigari, MD, Gaetano Gargiulo, MD, Fernando Picchio, MD, Maria C. Digilio, MD, Silvia Anaclerio, MD, Gianluca Oricchio, MD, Stephen P. Sanders, MD, and Roberto M. Di Donato, MD

Ann Thor Surg, 2006; 81: 968-75



Impact of DEL22q11, trisomy 21, and other genetic syndromes on surgical outcome of conotruncal heart defects

Guido Michielon, MD, Bruno Marino, MD, Gianluca Oricchio, MD, Maria Cristina Digilio, MD, Fiore Iorio, MD, Sergio Filippelli, MD, Silvia Placidi, MD, and Roberto M. Di Donato, MD

JTCVS 2009;138:565-570

Human Molecular Genetics, 1997, Vol. 6, No. 2 259–265

UFD1L, a developmentally expressed ubiquitination gene, is deleted in CATCH 22 syndrome

Antonio Pizzuti[1,2], Giuseppe Novelli[3], Antonia Ratti[1], Francesca Amati[2,3], Aldo Mari[2,3], Giuseppe Calabrese[4], Silvia Nicolis[5], Vincenzo Silani[6], Bruno Marino[7], Guglielmo Scarlato[1], Sergio Ottolenghi[5] and Bruno Dallapiccola[2,3,*]

Hum Molec Genet 1997; 6(2): 259-265

Ospedale Pediatrico
Bambino Gesù
ISTITUTO DI RICOVERO E CURA
A CARATTERE SCIENTIFICO

DELETION 22q11.2
Third International Meeting

Rome, Italy
June 7-8 , 2002

Pontificia Università Urbaniana
Auditorium "Giovanni Paolo II


AIdel22
Associazione Italiana delezione del Cromosoma 22
AIdel22


Lombardia
Friuli Venezia Giulia
Piemonte
Veneto
Emilia Romagna
Liguria
Marche
Toscana
Umbria
Abruzzo
Lazio
Puglia
Roma
Campania
Sardegna
Calabria
Sicilia
Referenti Regionali-AIdel22
Sede Nazionale AIdel22

Roma 3-5 luglio 2009

AIdel22: congressi italiani 2003-2012

2003-2004

Roma
17 Maggio
Istituto Caetani
"Aspetti clinici della Delezione del cromosoma 22"

Roma
13 Marzo
Ospedale Pediatrico Bambino Gesù
"Convegno Internazionale: la Delezione 22q11.2"

2005

Roma
26 Febbraio
Istituto Caetani
"L'apprendimento in situazione di svantaggio. Proposta di indagine sull'esperienza scolastica di ragazzi con del22"

Parma
27,28 Maggio
in collaborazione con AIEOP Comitato strategico e di Studio per le Immunodeficienze
"Protocollo diagnostico e terapeutico del22"

Torino
19 Novembre
In collaborazione con Ospedale Regina Margherita
"Dalla Sindrome di Di George alla Del 22 l'evoluzione di un approccio"

2006

Milano
26 Febbraio
Teatro Blu
"Aspetti diagnostici e terapeutici in età adulta"

Roma
25 Marzo
Ospedale Pediatrico Bambino Gesù
"Assistenza integrata per i pazienti affetti da malattia genetica complessa e disabilità congenita"

Strasburgo
7,8,9 Luglio
In collaborazione con Generation 22 e VCFS EF.Inc
"12°Annual International Scientific Meeting" The VeloCardio-Facial Sindrome Educational Foundation"

Roma
18 Novembre
Ospedale Pediatrico Bambino Gesù
"Al ritorno da Strasburgo 2006: un incontro con le famiglie italiane"

2007-2008

Firenze
31 Marzo
Ospedale Anna Meyer
"Convegno sugli aspetti comportamentali, neuro-psicologici e psichiatrici della sindrome del 22"

Bologna
12 Aprile
Istituto Clinico di Pediatria Preventiva e Neonatologia
"Approccio integrato al bambino e all'adolescente con Sindrome da Delezione del Cromosoma 22"

2009

Roma
3-5 Luglio
Hotel Pineta Palace
Aidel22 e VCFS EF.Inc.
"XVI International Scientific Meeting- The VeloCardio-Facial Sindrome Educational Foundation"

2011-2012

Napoli
26 Marzo
Istituto di Genetica e Biofisica C.N.R.
"Ricerca e Gestione clinica: un approccio unitario agli aspetti neuropsichiatrici e comportamentali nella del22"

Venezia
6 Ottobre
Ateneo Veneto
Presentazione Bilancio Sociale 2002-2012

Molecular Characterization of Chromosome 22 Deletions by Short Tandem Repeat Polymorphism (STRP) in Patients with Conotruncal Heart Defects

Simona Vittorini, Monica Sacchelli, Maria R. Iascone, Anita Collavoli, Simona Storti, Andrea Giusti, Giovanna Andreani, Nicoletta Botto, Andrea Biagini, Aldo Clerico

Laboratorio di Biologia Molecolare, IFC-CNR, Ospedale G. Pasquinucci, Massa, Italy

Clin Chem Lab Med 2001; 39: 1249-58

Safety and immunogenicity of measles–mumps–rubella vaccine in children with congenital immunodeficiency (DiGeorge syndrome)

Chiara Azzari*, Eleonora Gambineri, Massimo Resti, Maria Moriondo, Letizia Betti, Lucien Rojas Saldias, Anna M. G. Gelli, Alberto Vierucci

Department of Pediatrics, University of Florence, Pediatric Hospital A. Meyer, Via Luca Giordano, 13, 50132 Firenze, Italy

Vaccine 2005; 23: 1668-1671

Renzo Guerrini · Carla Marini

Genetic malformations of cortical development

Epilepsy, Neurophysiology and Neurogenetics Unit, Division of Child Neurology and Psychiatry, University of Pisa and Research Institute Stella Maris Foundation, Calambrone, Pisa, Italy

Exp Brai Res 2006; 173: 322-333

Expanding the phenotype of 22q11 deletion syndrome: the MURCS association

Vera Uliana[a], Nicola Giordano[b], Rossella Caselli[a], Filomena Tiziana Papa[a], Francesca Ariani[a], Claudio Marcocci[c], Elena Gianetti[c], Giuseppe Martini[b], Panagiotis Papakostas[b], Fabio Rollo[b], Ilaria Meloni[a], Francesca Mari[a], Manuela Priolo[d], Alessandra Renieri[a] and Ranuccio Nuti[b]

Medical Genetics, Department of Molecular Biology, University of Siena, Siena, Italy

Clin Dismorph 2008; 17: 13-7

Thyroid function and morphology in subjects with microdeletion of chromosome 22q11 (del(22)(q11))

Stefano Stagi*, Elisabetta Lapit, Eleonora Gambineri‡, Roberto Salti*, Maurizio Genuardit, Gloria Colarusso‡, Camilla Conti‡, Rita Jenuso§, Francesco Chiarelli¶, Chiara Azzari‡ and Maurizio de Martino‡

*Paediatric Endocrinology Unit, †Genetics and Molecular Medicine Unit, ‡Paediatric Immunology Unit, §Paediatric Radiology Unit, University of Florence, Anna Meyer Children's Hospital, Florence, and ¶Department of Pediatrics, University of Chieti, Chieti, Italy

Clin Endocrin 2010; 72: 839-844

Del22: Linee guida internazionali

Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome

Anne S. Bassett, MD,* Donna M. McDonald-McGinn, MS, CGC,* Koen Devriendt, MD, Maria Cristina Digilio, MD, Paula Goldenberg, MD, MSW, Alex Habel, MD, Bruno Marino, MD, Solveig Oskarsdottir, MD, PhD, Nicole Philip, MD, Kathleen Sullivan, MD, PhD, Ann Swillen, PhD, Jacob Vorstman, MD, PhD, and The International 22q11.2 Deletion Syndrome Consortium**

J Pediatr 2011;159:332-339

Table II. Recommended assessments for 22q11.2 deletion syndrome*

Assessment	At diagnosis	Infancy (0-12 months)	Preschool age (1-5 years)	School age (6-11 years)	Adolescence (12-18 years)	Adulthood (>18 years)
Ionized calcium, parathyroid hormone†	✓	✓	✓	✓	✓	✓
Thyrotropin (thyroid-stimulating hormone)†	✓		✓	✓	✓	✓
Complete blood cell count and differential (annual)	✓	✓	✓	✓	✓	✓
Immunologic evaluation‡	✓	✓§	✓§			
Ophthalmology	✓		✓			
Evaluate palate¶	✓	✓	✓			
Audiology	✓	✓	✓			✓
Cervical spine (>age 4 years)			✓‖			
Scoliosis examination	✓		✓		✓	
Dental evaluation			✓	✓	✓	✓
Renal ultrasound	✓					
Electrocardiogram	✓					✓
Echocardiogram	✓					
Development**	✓	✓	✓			
School performance				✓	✓	
Socialization/functioning	✓	✓	✓	✓	✓	✓
Psychiatric/emotional/behavioral††	✓		✓	✓	✓	✓
Systems review	✓	✓	✓	✓	✓	✓
Deletion studies of parents	✓					
Genetic counseling‡‡	✓				✓	✓
Gynecologic and contraceptive services					✓	✓

Del22: Linee guida internazionali

"LA PAROLA del 22"
Roma, 24 e 25 maggio 2013
Auditorium San Paolo
Ospedale Pediatrico Bambino Gesù IRCCS
Aidel22 Bambino Gesù

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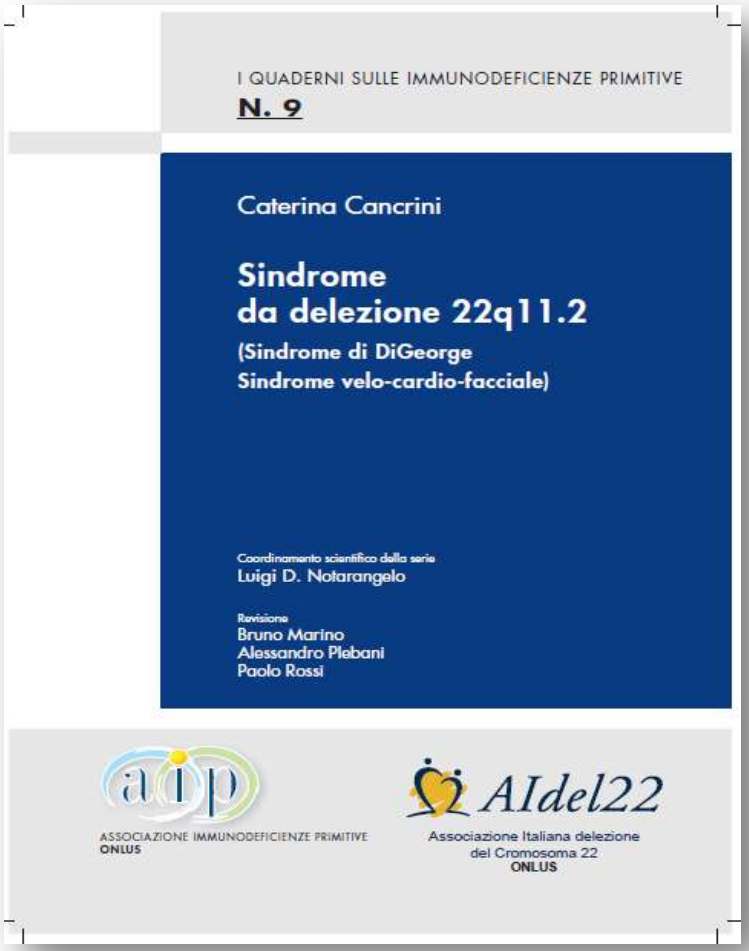
Immunologic evaluation‡	✓	✓§	✓§			
Ophthalmology	✓		✓			
Evaluate palate¶	✓	✓	✓			
Audiology	✓	✓	✓			✓
Cervical spine (>age 4 years)			✓‖			
Scoliosis examination	✓		✓		✓	
Dental evaluation			✓	✓	✓	✓
Renal ultrasound	✓					
Electrocardiogram	✓					✓
Echocardiogram	✓					
Development**	✓	✓	✓			
School performance				✓	✓	
Socialization/functioning	✓	✓	✓	✓	✓	✓
Psychiatric/emotional/behavioral††	✓		✓	✓	✓	✓
Systems review	✓	✓	✓	✓	✓	✓
Deletion studies of parents	✓					
Genetic counseling‡‡	✓				✓	✓
Gynecologic and contraceptive services					✓	✓

Del22: Studio multicentrico italiano



ipinet
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Associazione Italiana
Ematologia Oncologia Pediatrica

Del22: Progetto Adolescente-Adulto

Policlinico Umberto I – Università La Sapienza, Roma

- Cardiologo
- Genetista clinico
- Psichiatra
- Endocrinologo
- Neurologo
- Immunologo
- Gastroenterologo
- Otorinolaringoiatra
- Urologo/Ginecologo
- Ortopedico
- Oculista
- Dermatologo